EGFR gene

epidermal growth factor receptor

Normal Function

The *EGFR* gene provides instructions for making a receptor protein called the epidermal growth factor receptor, which spans the cell membrane so that one end of the protein remains inside the cell and the other end projects from the outer surface of the cell. This positioning allows the receptor to attach (bind) to other proteins, called ligands, outside the cell and to receive signals that help the cell respond to its environment. Ligands and receptors fit together like keys into locks. Epidermal growth factor receptor binds to at least seven different ligands. The binding of a ligand to an epidermal growth factor receptor allows the receptor to attach to a nearby receptor protein (dimerize), turning on (activating) the receptor complex. As a result, signaling pathways within the cell are triggered that promote cell growth and division (proliferation) and cell survival.

Health Conditions Related to Genetic Changes

lung cancer

At least eight mutations in the *EGFR* gene have been associated with lung cancer, a disease in which certain cells in the lung become abnormal and multiply uncontrollably to form a tumor. Nearly all these *EGFR* gene mutations occur during a person's lifetime (somatic) and are only present in cancer cells. Other genetic, environmental, and lifestyle factors also contribute to a person's cancer risk; in lung cancer, the greatest risk factor is being a long-term tobacco smoker.

Somatic mutations in the *EGFR* gene most often occur in a type of lung cancer called non-small cell lung cancer, specifically a form called adenocarcinoma. These mutations are most common in people with the disease who have never smoked. Somatic *EGFR* gene mutations occur more frequently in Asian populations with lung cancer than in affected white populations, occurring in 30 to 40 percent of affected Asians compared to 10 to 15 percent of whites with lung cancer.

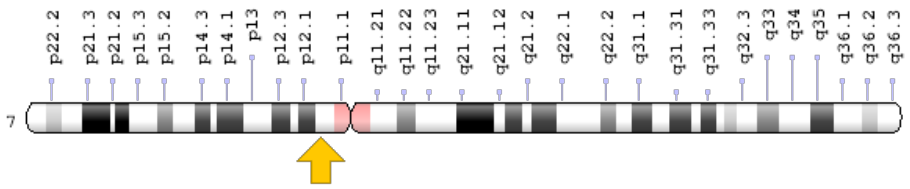
Most of the somatic *EGFR* gene mutations that are associated with lung cancer delete genetic material in a part of the gene known as exon 19 or change DNA building blocks (nucleotides) in another region called exon 21. These gene changes result in a receptor protein that is constantly turned on (constitutively activated), even when it is not bound to a ligand. As a result, cells are signaled to constantly proliferate and survive, leading to tumor formation. When these gene changes occur in cells in the lungs, lung cancer develops.

Lung cancers with *EGFR* gene mutations tend to respond to treatments that target the overactive signaling pathways that allow cancer cells to constantly grow and divide.

Chromosomal Location

Cytogenetic Location: 7p11.2, which is the short (p) arm of chromosome 7 at position 11.2

Molecular Location: base pairs 55,019,032 to 55,207,338 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cell growth inhibiting protein 40
- cell proliferation-inducing protein 61
- erb-b2 receptor tyrosine kinase 1
- ERBB
- ERBB1
- HER1
- mENA
- NISBD2
- PIG61
- proto-oncogene c-ErbB-1
- receptor tyrosine-protein kinase erbB-1

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EGFR%5BTI%5D%29+OR+%28epidermal+growth+factor+receptor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- EPIDERMAL GROWTH FACTOR RECEPTOR
  http://omim.org/entry/131550

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EGFR.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EGFR%5Bgene%5D
- HGNC Gene Family: Erb-b2 receptor tyrosine kinases
  http://www.genenames.org/cgi-bin/genefamilies/set/1096
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3236
- IUPHAR Database
  http://www.guidetopharmacology.org/GRAC/ObjectDisplayForward?objectId=1797
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1956
- UniProt
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